SEQUENCE LISTING

<110> Omary, M. Bishr
      Ku, Nam-Om

<120> Keratin 8 and 18 mutations are Risk
      Factors for Developing Liver Disease of Multiple Etiologies

<130> STAN-297WO

<150> 60/462,989
<151> 2003-04-14

<160> 4

<170> FastSEQ for Windows Version 4.0

<210> 1
<211> 1485
<212> DNA
<213> H. sapiens

<220>
<221> CDS
<222> (119)...(1408)

<400> 1
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ccgccaccgt cgtccgcaaa gcctgagtcc tgtcctttct ctctccccgg acagcatg   118
agc ttc acc act cgc tcc acc ttc tcc acc aac tac cgg tcc ctg ggc   166
Ser Phe Thr Thr Arg Ser Thr Phe Ser Thr Asn Tyr Arg Ser Leu Gly
1   5   10   15

tct gtc cag gcg ccc agc tac ggc gcc cgg ccg gtc agc agc gcg gcc   214
Ser Val Gln Ala Pro Ser Tyr Gly Ala Arg Pro Val Ser Ser Ala Ala
            20   25   30

agc gtc tat gca ggc gct ggg ggc tct ggt tcc cgg atc tcc gtg tcc   262
Ser Val Tyr Ala Gly Ala Gly Gly Ser Gly Ser Arg Ile Ser Val Ser
            35   40   45

cgc tcc acc agc ttc agg ggc ggc atg ggg tcc ggg ggc ctg gcc acc   310
Arg Ser Thr Ser Phe Arg Gly Gly Met Gly Ser Gly Gly Leu Ala Thr
        50   55   60

ggg ata gcc ggg ggt ctg gca gga atg gga ggc atc cag aac gag aag   358
Gly Ile Ala Gly Gly Leu Ala Gly Met Gly Gly Ile Gln Asn Glu Lys
65   70   75   80

gag acc atg caa agc ctg aac gac cgc ctg gcc tct tac ctg gac aga   406
Glu Thr Met Gln Ser Leu Asn Asp Arg Leu Ala Ser Tyr Leu Asp Arg
                85   90   95

gtg agg agc ctg gag acc gag aac cgg agg ctg gag agc aaa atc cgg   454
Val Arg Ser Leu Glu Thr Glu Asn Arg Arg Leu Glu Ser Lys Ile Arg
            100   105   110

gag cac ttg gag aag aag gga ccc cag gtc aga gac tgg agc cat tac   502
Glu His Leu Glu Lys Lys Gly Pro Gln Val Arg Asp Trp Ser His Tyr
            115   120   125

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ttc aag atc atc gag gac ctg agg gct cag atc ttc gca aat act gtg      550
Phe Lys Ile Ile Glu Asp Leu Arg Ala Gln Ile Phe Ala Asn Thr Val
    130             135                 140

gac aat gcc cgc atc gtt ctg cag att gac aat gcc cgt ctt gct gct      598
Asp Asn Ala Arg Ile Val Leu Gln Ile Asp Asn Ala Arg Leu Ala Ala
    145             150                 155             160

gat gac ttt aga gtc aag tat gag aca gag ctg gcc atg cgc cag tct      646
Asp Asp Phe Arg Val Lys Tyr Glu Thr Glu Leu Ala Met Arg Gln Ser
            165             170                 175

gtg gag aac gac atc cat ggg ctc cgc aag gtc att gat gac acc aat      694
Val Glu Asn Asp Ile His Gly Leu Arg Lys Val Ile Asp Asp Thr Asn
        180             185                 190

atc aca cga ctg cag ctg gag aca gag atc gag gct ctc aag gag gag      742
Ile Thr Arg Leu Gln Leu Glu Thr Glu Ile Glu Ala Leu Lys Glu Glu
            195             200                 205

ctg ctc ttc atg aag aag aac cac gaa gag gaa gta aaa ggc cta caa      790
Leu Leu Phe Met Lys Lys Asn His Glu Glu Glu Val Lys Gly Leu Gln
    210             215                 220

gcc cag att gcc agc tct ggg ttg acc gtg gag gta gat gcc ccc aaa      838
Ala Gln Ile Ala Ser Ser Gly Leu Thr Val Glu Val Asp Ala Pro Lys
225             230                 235             240

tct cag gac ctc gcc aag atc atg gca gac atc cgg gcc caa tat gac      886
Ser Gln Asp Leu Ala Lys Ile Met Ala Asp Ile Arg Ala Gln Tyr Asp
            245             250                 255

gag ctg gct cgg aag aac cga gag gag cta gac aag tac tgg tct cag      934
Glu Leu Ala Arg Lys Asn Arg Glu Glu Leu Asp Lys Tyr Trp Ser Gln
            260             265                 270

cag att gag gag agc acc aca gtg gtc acc aca cag tct gct gag gtt      982
Gln Ile Glu Glu Ser Thr Thr Val Val Thr Thr Gln Ser Ala Glu Val
    275             280                 285

gga gct gct gag acg acg ctc aca gag ctg aga cgt aca gtc cag tcc      1030
Gly Ala Ala Glu Thr Thr Leu Thr Glu Leu Arg Arg Thr Val Gln Ser
    290             295                 300

ttg gag atc gac ctg gac tcc atg aga aat ctg aag gcc agc ttg gag      1078
Leu Glu Ile Asp Leu Asp Ser Met Arg Asn Leu Lys Ala Ser Leu Glu
305             310                 315             320

aac agc ctg agg gag gtg gag gcc cgc tac gcc cta cag atg gag cag      1126
Asn Ser Leu Arg Glu Val Glu Ala Arg Tyr Ala Leu Gln Met Glu Gln
            325             330                 335

ctc aac ggg atc ctg ctg cac ctt gag tca gag ctg gca cag acc cgg      1174
Leu Asn Gly Ile Leu Leu His Leu Glu Ser Glu Leu Ala Gln Thr Arg
            340             345                 350

gca gag gga cag cgc cag gcc cag gag tat gag gcc ctg ctg aac atc      1222
Ala Glu Gly Gln Arg Gln Ala Gln Glu Tyr Glu Ala Leu Leu Asn Ile
            355             360                 365
```

```
aag gtc aag ctg gag gct gag atc gcc acc tac cgc cgc ctg ctg gaa      1270
Lys Val Lys Leu Glu Ala Glu Ile Ala Thr Tyr Arg Arg Leu Leu Glu
        370             375             380

gat ggc gag gac ttt aat ctt ggt gat gcc ttg gac agc agc aac tcc      1318
Asp Gly Glu Asp Phe Asn Leu Gly Asp Ala Leu Asp Ser Ser Asn Ser
385             390             395             400

atg caa acc atc caa aag acc acc acc cgc cgg ata gtg gat ggc aaa      1366
Met Gln Thr Ile Gln Lys Thr Thr Thr Arg Arg Ile Val Asp Gly Lys
                405             410             415

gtg gtg tct gag acc aat gac acc aaa gtt ctg agg cat taa            1408
Val Val Ser Glu Thr Asn Asp Thr Lys Val Leu Arg His  *
        420             425

    gccagcagaa gcagggtacc ctttggggag caggaggcca ataaaaagtt cagagttcaa     1468
    aaaaaaaaaa aaaaaaa                                                    1485


<210> 2
<211> 429
<212> PRT
<213> H. sapiens

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Ser Phe Thr Thr Arg Ser Thr Phe Ser Thr Asn Tyr Arg Ser Leu Gly
1               5                   10                  15
Ser Val Gln Ala Pro Ser Tyr Gly Ala Arg Pro Val Ser Ser Ala Ala
            20                  25                  30
Ser Val Tyr Ala Gly Ala Gly Gly Ser Gly Ser Arg Ile Ser Val Ser
            35                  40                  45
Arg Ser Thr Ser Phe Arg Gly Gly Met Gly Ser Gly Gly Leu Ala Thr
        50                  55                  60
Gly Ile Ala Gly Gly Leu Ala Gly Met Gly Gly Ile Gln Asn Glu Lys
65                  70                  75                  80
Glu Thr Met Gln Ser Leu Asn Asp Arg Leu Ala Ser Tyr Leu Asp Arg
                85                  90                  95
Val Arg Ser Leu Glu Thr Glu Asn Arg Arg Leu Glu Ser Lys Ile Arg
            100                 105                 110
Glu His Leu Glu Lys Lys Gly Pro Gln Val Arg Asp Trp Ser His Tyr
        115                 120                 125
Phe Lys Ile Ile Glu Asp Leu Arg Ala Gln Ile Phe Ala Asn Thr Val
    130                 135                 140
Asp Asn Ala Arg Ile Val Leu Gln Ile Asp Asn Ala Arg Leu Ala Ala
145                 150                 155                 160
Asp Asp Phe Arg Val Lys Tyr Glu Thr Glu Leu Ala Met Arg Gln Ser
                165                 170                 175
Val Glu Asn Asp Ile His Gly Leu Arg Lys Val Ile Asp Asp Thr Asn
            180                 185                 190
Ile Thr Arg Leu Gln Leu Glu Thr Glu Ile Glu Ala Leu Lys Glu Glu
        195                 200                 205
Leu Leu Phe Met Lys Lys Asn His Glu Glu Glu Val Lys Gly Leu Gln
    210                 215                 220
Ala Gln Ile Ala Ser Ser Gly Leu Thr Val Glu Val Asp Ala Pro Lys
225                 230                 235                 240
Ser Gln Asp Leu Ala Lys Ile Met Ala Asp Ile Arg Ala Gln Tyr Asp
                245                 250                 255
Glu Leu Ala Arg Lys Asn Arg Glu Glu Leu Asp Lys Tyr Trp Ser Gln
            260                 265                 270
Gln Ile Glu Glu Ser Thr Thr Val Val Thr Thr Gln Ser Ala Glu Val
        275                 280                 285
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```
Gly Ala Ala Glu Thr Thr Leu Thr Glu Leu Arg Arg Thr Val Gln Ser
    290                 295                 300
Leu Glu Ile Asp Leu Asp Ser Met Arg Asn Leu Lys Ala Ser Leu Glu
305                 310                 315                 320
Asn Ser Leu Arg Glu Val Glu Ala Arg Tyr Ala Leu Gln Met Glu Gln
            325                 330                 335
Leu Asn Gly Ile Leu Leu His Leu Glu Ser Glu Leu Ala Gln Thr Arg
            340                 345                 350
Ala Glu Gly Gln Arg Gln Ala Gln Glu Tyr Glu Ala Leu Leu Asn Ile
        355                 360                 365
Lys Val Lys Leu Glu Ala Glu Ile Ala Thr Tyr Arg Arg Leu Leu Glu
    370                 375                 380
Asp Gly Glu Asp Phe Asn Leu Gly Asp Ala Leu Asp Ser Ser Asn Ser
385                 390                 395                 400
Met Gln Thr Ile Gln Lys Thr Thr Thr Arg Arg Ile Val Asp Gly Lys
            405                 410                 415
Val Val Ser Glu Thr Asn Asp Thr Lys Val Leu Arg His
            420                 425
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<210> 3
<211> 1752
<212> DNA
<213> H. sapiens

<220>
<221> CDS
<222> (63)...(1511)

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tg tcc atc agg gtg acc cag aag tcc tac aag gtg tcc acc tct ggc      107
   Ser Ile Arg Val Thr Gln Lys Ser Tyr Lys Val Ser Thr Ser Gly
   1               5                   10                  15

ccc cgg gcc ttc agc agc cgc tcc tac acg agt ggg ccc ggt tcc cgc    155
Pro Arg Ala Phe Ser Ser Arg Ser Tyr Thr Ser Gly Pro Gly Ser Arg
                20                  25                  30

atc agc tcc tcg agc ttc tcc cga gtg ggc agc agc aac ttt cgc ggt    203
Ile Ser Ser Ser Ser Phe Ser Arg Val Gly Ser Ser Asn Phe Arg Gly
            35                  40                  45

ggc ctg ggc ggc ggc tat ggt ggg gcc agc ggc atg gga ggc atc acc    251
Gly Leu Gly Gly Gly Tyr Gly Gly Ala Ser Gly Met Gly Gly Ile Thr
        50                  55                  60

gca gtt acg gtc aac cag agc ctg ctg agc ccc ctt gtc ctg gag gtg    299
Ala Val Thr Val Asn Gln Ser Leu Leu Ser Pro Leu Val Leu Glu Val
    65                  70                  75

gac ccc aac atc cag gcc gtg cgc acc cag gag aag gag cag atc aag    347
Asp Pro Asn Ile Gln Ala Val Arg Thr Gln Glu Lys Glu Gln Ile Lys
80                  85                  90                  95

acc ctc aac aac aag ttt gcc tcc ttc ata gac aag gta cgg ttc ctg    395
Thr Leu Asn Asn Lys Phe Ala Ser Phe Ile Asp Lys Val Arg Phe Leu
                100                 105                 110

gag cag cag aac aag atg ctg gag acc aag tgg agc ctc ctg cag cag    443
Glu Gln Gln Asn Lys Met Leu Glu Thr Lys Trp Ser Leu Leu Gln Gln
            115                 120                 125
```

```
cag aag acg gct cga agc aac atg gac aac atg ttc gag agc tac atc    491
Gln Lys Thr Ala Arg Ser Asn Met Asp Asn Met Phe Glu Ser Tyr Ile
        130                 135                 140

aac aac ctt agg cgg cag ctg gag act ctg ggc cag gag aag ctg aag    539
Asn Asn Leu Arg Arg Gln Leu Glu Thr Leu Gly Gln Glu Lys Leu Lys
        145                 150                 155

ctg gag gcg gag ctt ggc aac atg cag ggg ctg gtg gag gac ttc aag    587
Leu Glu Ala Glu Leu Gly Asn Met Gln Gly Leu Val Glu Asp Phe Lys
160                 165                 170                 175

aac aag tat gag gat gag atc aat aag cgt aca gag atg gag aac gaa    635
Asn Lys Tyr Glu Asp Glu Ile Asn Lys Arg Thr Glu Met Glu Asn Glu
                180                 185                 190

ttt gtc ctc atc aag aag gat gtg gat gaa gct tac atg aac aag gta    683
Phe Val Leu Ile Lys Lys Asp Val Asp Glu Ala Tyr Met Asn Lys Val
                195                 200                 205

gag ctg gag tct cgc ctg gaa ggg ctg acc gac gag atc aac ttc ctc    731
Glu Leu Glu Ser Arg Leu Glu Gly Leu Thr Asp Glu Ile Asn Phe Leu
        210                 215                 220

agg cag cta tat gaa gag gag atc cgg gag ctg cag tcc cag atc tcg    779
Arg Gln Leu Tyr Glu Glu Glu Ile Arg Glu Leu Gln Ser Gln Ile Ser
        225                 230                 235

gac aca tct gtg gtg ctg tcc atg gac aac agc cgc tcc ctg gac atg    827
Asp Thr Ser Val Val Leu Ser Met Asp Asn Ser Arg Ser Leu Asp Met
240                 245                 250                 255

gac agc atc att gct gag gtc aag gca cag tac gag gat att gcc aac    875
Asp Ser Ile Ile Ala Glu Val Lys Ala Gln Tyr Glu Asp Ile Ala Asn
                260                 265                 270

cgc agc cgg gct gag gct gag agc atg tac cag atc aag tat gag gag    923
Arg Ser Arg Ala Glu Ala Glu Ser Met Tyr Gln Ile Lys Tyr Glu Glu
                275                 280                 285

ctg cag agc ctg gct ggg aag cac ggg gat gac ctg cgg cgc aca aag    971
Leu Gln Ser Leu Ala Gly Lys His Gly Asp Asp Leu Arg Arg Thr Lys
        290                 295                 300

act gag atc tct gag atg aac cgg aac atc agc cgg ctc cag gct gag   1019
Thr Glu Ile Ser Glu Met Asn Arg Asn Ile Ser Arg Leu Gln Ala Glu
        305                 310                 315

att gag ggc ctc aaa ggc cag agg gct tcc ctg gag gcc gcc att gca   1067
Ile Glu Gly Leu Lys Gly Gln Arg Ala Ser Leu Glu Ala Ala Ile Ala
320                 325                 330                 335

gat gcc gag cag cgt gga gag ctg gcc att aag gat gcc aac gcc aag   1115
Asp Ala Glu Gln Arg Gly Glu Leu Ala Ile Lys Asp Ala Asn Ala Lys
                340                 345                 350

ttg tcc gag ctg gag gcc gcc ctg cag cgg gcc aag cag gac atg gcg   1163
Leu Ser Glu Leu Glu Ala Ala Leu Gln Arg Ala Lys Gln Asp Met Ala
        355                 360                 365
```

```
cgg cag ctg cgt gag tac cag gag ctg atg aac gtc aag ctg gcc ctg       1211
Arg Gln Leu Arg Glu Tyr Gln Glu Leu Met Asn Val Lys Leu Ala Leu
        370             375             380

gac atc gag atc gcc acc tac agg aag ctg ctg gag ggc gag gag agc       1259
Asp Ile Glu Ile Ala Thr Tyr Arg Lys Leu Leu Glu Gly Glu Glu Ser
    385             390             395

cgg ctg gag tct ggg atg cag aac atg agt att cat acg aag acc acc       1307
Arg Leu Glu Ser Gly Met Gln Asn Met Ser Ile His Thr Lys Thr Thr
400             405             410             415

agc ggc tat gca ggt ggt ctg agc tcg gcc tat ggg ggc ctc aca agc       1355
Ser Gly Tyr Ala Gly Gly Leu Ser Ser Ala Tyr Gly Gly Leu Thr Ser
            420             425             430

ccc ggc ctc agc tac agc ctg ggc tcc agc ttt ggc tct ggc gcg ggc       1403
Pro Gly Leu Ser Tyr Ser Leu Gly Ser Ser Phe Gly Ser Gly Ala Gly
            435             440             445

tcc agc tcc ttc agc cgc acc agc tcc tcc agg gcc gtg gtt gtg aag       1451
Ser Ser Ser Phe Ser Arg Thr Ser Ser Ser Arg Ala Val Val Val Lys
            450             455             460

aag atc gag aca cgt gat ggg aag ctg gtg tct gag tcc tct gac gtc       1499
Lys Ile Glu Thr Arg Asp Gly Lys Leu Val Ser Glu Ser Ser Asp Val
    465             470             475

ctg ccc aag tga acagctgcgg cagcccctcc cagcctaccc ctcctgcgct          1551
Leu Pro Lys *
480
```

```
gccccagagc ctgggaagga ggccgctatg cagggtagca ctgggaacag gagacccacc      1611
tgaggctcag ccctagccct cagcccacct ggggagttta ctacctgggg accccccttg      1671
cccatgcctc cagctacaaa acaattcaat tgcttttttt ttttggtcca aaataaaacc      1731
tcagctagct ctgccaaacc c                                                1752
```

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<210> 4
<211> 482
<212> PRT
<213> H. sapiens

<400> 4
Ser Ile Arg Val Thr Gln Lys Ser Tyr Lys Val Ser Thr Ser Gly Pro
 1               5              10              15
Arg Ala Phe Ser Ser Arg Ser Tyr Thr Ser Gly Pro Gly Ser Arg Ile
            20              25              30
Ser Ser Ser Ser Phe Ser Arg Val Gly Ser Ser Asn Phe Arg Gly Gly
        35              40              45
Leu Gly Gly Gly Tyr Gly Gly Ala Ser Gly Met Gly Gly Ile Thr Ala
    50              55              60
Val Thr Val Asn Gln Ser Leu Leu Ser Pro Leu Val Leu Glu Val Asp
65              70              75              80
Pro Asn Ile Gln Ala Val Arg Thr Gln Glu Lys Glu Gln Ile Lys Thr
            85              90              95
Leu Asn Asn Lys Phe Ala Ser Phe Ile Asp Lys Val Arg Phe Leu Glu
            100             105             110
Gln Gln Asn Lys Met Leu Glu Thr Lys Trp Ser Leu Leu Gln Gln Gln
        115             120             125
Lys Thr Ala Arg Ser Asn Met Asp Asn Met Phe Glu Ser Tyr Ile Asn
    130             135             140
```

```
Asn Leu Arg Arg Gln Leu Glu Thr Leu Gly Gln Glu Lys Leu Lys Leu
145                 150                 155                 160
Glu Ala Glu Leu Gly Asn Met Gln Gly Leu Val Glu Asp Phe Lys Asn
                165                 170                 175
Lys Tyr Glu Asp Glu Ile Asn Lys Arg Thr Glu Met Glu Asn Glu Phe
                180                 185                 190
Val Leu Ile Lys Lys Asp Val Asp Glu Ala Tyr Met Asn Lys Val Glu
            195                 200                 205
Leu Glu Ser Arg Leu Glu Gly Leu Thr Asp Glu Ile Asn Phe Leu Arg
        210                 215                 220
Gln Leu Tyr Glu Glu Glu Ile Arg Glu Leu. Gln Ser Gln Ile Ser Asp
225                 230                 235                 240
Thr Ser Val Val Leu Ser Met Asp Asn Ser Arg Ser Leu Asp Met Asp
                245                 250                 255
Ser Ile Ile Ala Glu Val Lys Ala Gln Tyr Glu Asp Ile Ala Asn Arg
            260                 265                 270
Ser Arg Ala Glu Ala Glu Ser Met Tyr Gln Ile Lys Tyr Glu Glu Leu
        275                 280                 285
Gln Ser Leu Ala Gly Lys His Gly Asp Asp Leu Arg Arg Thr Lys Thr
    290                 295                 300
Glu Ile Ser Glu Met Asn Arg Asn Ile Ser Arg Leu Gln Ala Glu Ile
305                 310                 315                 320
Glu Gly Leu Lys Gly Gln Arg Ala Ser Leu Glu Ala Ala Ile Ala Asp
            325                 330                 335
Ala Glu Gln Arg Gly Glu Leu Ala Ile Lys Asp Ala Asn Ala Lys Leu
            340                 345                 350
Ser Glu Leu Glu Ala Ala Leu Gln Arg Ala Lys Gln Asp Met Ala Arg
        355                 360                 365
Gln Leu Arg Glu Tyr Gln Glu Leu Met Asn Val Lys Leu Ala Leu Asp
370                 375                 380
Ile Glu Ile Ala Thr Tyr Arg Lys Leu Leu Glu Gly Glu Glu Ser Arg
.385                390                 395                 400
Leu Glu Ser Gly Met Gln Asn Met Ser Ile His Thr Lys Thr Thr Ser
                405                 410                 415
Gly Tyr Ala Gly Gly Leu Ser Ser Ala Tyr Gly Gly Leu Thr Ser Pro
            420                 425                 430
Gly Leu Ser Tyr Ser Leu Gly Ser Ser Phe Gly Ser Gly Ala Gly Ser
    435                 440                 445
Ser Ser Phe Ser Arg Thr Ser Ser Arg Ala Val Val Val Lys Lys
    450                 455                 460
Ile Glu Thr Arg Asp Gly Lys Leu Val Ser Glu Ser Ser Asp Val Leu
465                 470                 475                 480
Pro Lys
```